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Case Report

Crouzon Syndrome: Classical Presentation of A Rare Entity

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Abstract

Crouzon syndrome is the most common syndrome among the craniosynostosis group. Crouzon syndrome accounts for about 4.8% of all of them. It commonly has autosomal dominant inheritance with complete penetrance and variable expressivity from subtle to severe forms and characterized by craniosynostosis, exophthalmos, and hypoplastic maxilla with relative mandibular prognathism. Mutation of the fibroblast growth factor receptor-2 (FGFR2) gene is responsible for the occurrence of this rare genetic disorder.

Keywords: Craniosynostosis; Exophthalmos; FGFR-2

Introduction

Crouzon's syndrome (CS) is an autosomal dominant disorder, described by French neurosurgeon Octav Crouzan in 1912 [1]. It is caused by mutation in Fibroblast Growth Factor Receptor 2 (FGFR2) genes [2].This disease is characterized by premature synostosis of coronal and sagittal sutures. Prevalence rate of this syndromes is approximately 1 in every 25,000 live births. It is characterized by a triad of skull deformities, facial anomalies and exophthalmos. In our case, the patient had hypoplastic maxilla, prognathic mandible and mild exophthalmos.

Case Report

A 14-year old male patient was referred to the Department of Oral Medicine and Radiology for radiological evaluation of suspected syndromic involvement. Although the patient's father gave history that the patients suffers from frequent headaches, the patient appeared mentally alert and responsive. The family history was non-contributory. General examination revealed small built, mild frontal bossing, irregularly shaped cranial vault, sparse hair, mild hypertelorism, small ears (Figure 1), a deficient maxilla and prognathic mandible (Figure 2). Intra oral examination revealed grossly decayed upper and lower teeth and generalized spacing (Figure 3). A panoramic radiograph of the jaws confirmed the poor dental status of the patient. It also showed multiple radicular cysts and single rooted teeth (Figure 4). A lateral cephalogram showed a deficient maxilla, leading to mid face deficiency and slight mandibular prognathism (Figure 5). A PA skull view was diagnostic, showing the characteristic copper-plate beaten appearance of Crouzon Syndrome (Figure 6).

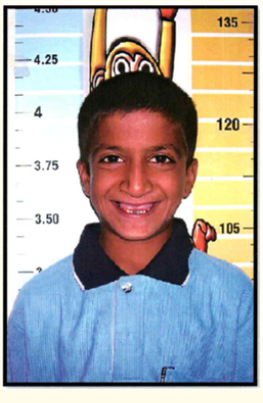

Figure 1: Small stature of patient.

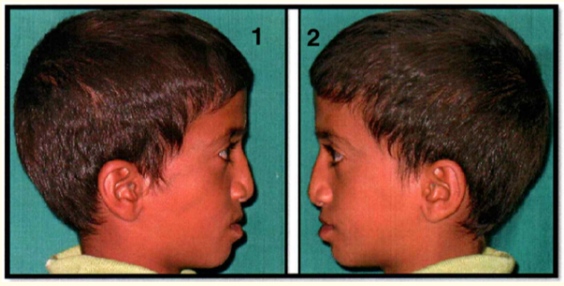

Figure 2: Right profile (1) and left profile (2) showing deficient mid face.

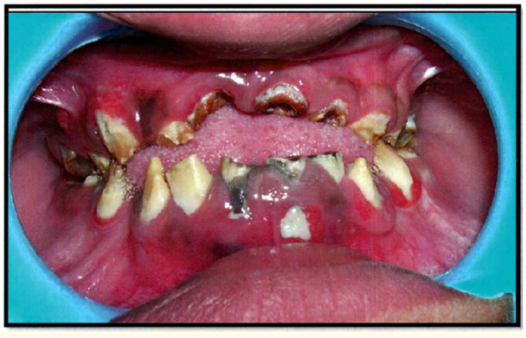

Figure 3: Poor dental status.

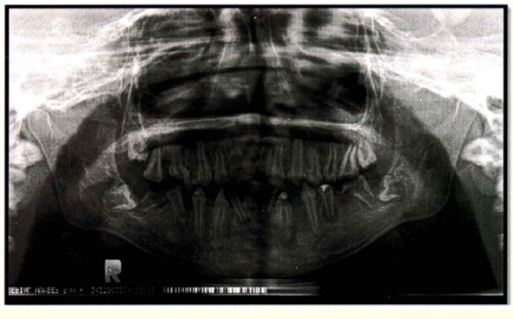

Figure 4: Panoramic radiograph of the jaws showing multiple radicular cysts and single rooted teeth.

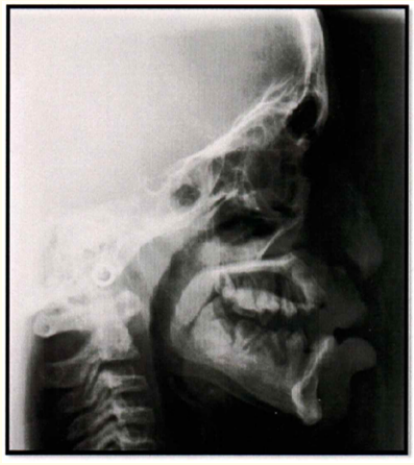

Figure 5: Lateral cephalogram showing deficient mid-face and prognathic mandible.

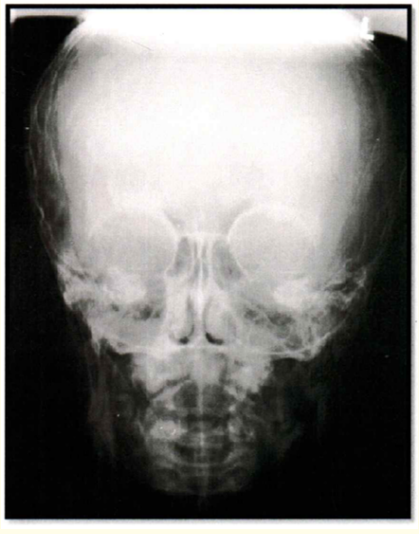

Figure 6: PA skull view showing 'copper beaten' appearance and maxillary sinus hypoplasia.

Discussion

CS is an autosomal dominant disorder with complete penetrance and variable expressivity [3,4]. It is characterized by a

triad of skull deformities, facial anomalies and exophthalmos [5]. There is no gender or sex predilection [6]. Other clinical features include hypertelorism, strabismus, beaked nose, short upper lip, maxillary hypoplasia and mandibular prognathism with no digital abnormalities [7,8]. In our case the patient had hypertelorism with mild exophthalmos, short upper lip, flattened nose, maxillary hypoplasia and prognathic mandible.

Diagnosis is based on clinical and radiographic findings. Radiographic findings include obliterated sutures, hypoplastic maxilla with shallow orbits, shortened cranial fossa, enlarged hypophyseal cavity and small paranasal sinuses [9]. In our case, Orthopantomogram, Lateral Cephalogram and PA Skull were advised to the patient. The radiographic examination revealed hypoplastic maxilla, prognathic mandible, copper beaten appearance of skull and bilateral maxillary sinus hypoplasia.

CS must be differentiated from simple craniosynostosis and other syndromic craniosynostosis. Differential diagnosis for other syndromic craniosynostosis includes Pfeiffer's syndrome, Apert syndrome, Saethre-Chotzen syndrome, Carpenter syndrome and Jackson-Weiss syndrome. CS can be differentiated from them on the basis of lack of involvement of hand, and/or feet.

Management of CS is multidisciplinary and early diagnosis is important. In the first year of life, it is preferred to release synostotic sutures of skull to allow adequate cranial volume thus allowing the brain to grow and expand. Skull reshaping may have to be repeated as the child grows to give best possible results [10,11].

Prognosis depends on the severity of malformations and the timing of intervention. Innovations in craniofacial surgery have enabled patients to achieve their full potential for intellectual growth, physical competence and social interactions.

Conclusion

Management of craniofacial deformities often requires multidisciplinary team involvement. Dental professionals should have sufficient knowledge of syndromes associated with dysmorphic faces to detect patients who are unaware of their condition, so they may be identified and sent for early investigations and management as required to prevent complications due to late diagnosis.

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